Opis choroby *

Definicja

A rare genetic disorder of lipid metabolism characterized by neonatal to childhood onset of impaired absorption of dietary fat with greasy/oily and voluminous stools, but normal growth and development. Decreased levels of fecal elastase, as well as low serum levels of the fat-soluble vitamins A, D, and E, have been reported.

Dane

Klasyfikacja
Choroba

Synonimy
Pancreatic triglyceride lipase deficiency
Niedobór trzustkowej lipazy trójglicerydów

Kod ORPHA
309031

Kod OMIM
614338

Kod ICD10
K90.3

Kod ICD11
5C62

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*Źródło

orphanet